GEPHE SUMMARY

Gephebase Gene
BMP15 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^BMP15^#gephebase-summary-title)

GepheID
GP00002157

Entry Status
Published

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Physiology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Physiology^#gephebase-summary-title)

Trait
Fertility (increased ovulation rate) (https://www.gephebase.org/search-criteria?/and+Trait=^Fertility (increased ovulation rate)^#gephebase-summary-title)

Trait State in Taxon A
Ovis aries

Trait State in Taxon B
Ovis aries - Belclare - High fertility in heterozygotes

Ancestral State
Taxon A

Taxonomic Status
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)

Taxon A

Latin Name
Ovis aries (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Ovis aries^#gephebase-summary-title)

Common Name
sheep

Synonyms
Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758

Rank
species

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Artiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Ovis

Parent
Ovis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9935)

NCBI Taxonomy ID
9940 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9940)

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Ovis aries (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Ovis aries^#gephebase-summary-title)

Common Name
sheep

Synonyms
Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758

Rank
species

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Artiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Ovis

Parent
Ovis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9935)

NCBI Taxonomy ID
9940 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9940)

is Taxon B an Infraspecies?
Yes

Taxon B Description
Ovis aries - Belclare ; Irish Cambridge

GENOTYPIC CHANGE

Generic Gene Name
Bmp15

Synonyms
Bmp-15; C86824; C87336; GDF-9B; AU015375; AU018861; AU021453; Gdf9b

String
10090.ENSMUSP00000024049 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000024049)

Sequence Similarities
Belongs to the TGF-beta family.

GO - Molecular Function
GO:0005125 : cytokine activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005125)
GO:0008083 : growth factor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008083)
GO:0005160 : transforming growth factor beta receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005160)

UniProtKB Mus musculus
Q9Z0L4 (http://www.uniprot.org/uniprot/Q9Z0L4)

GenebankID or UniProtKB
AHB23439 (https://www.ncbi.nlm.nih.gov/nuccore/AHB23439)

GO - Biological Process

GO:0045893 : positive regulation of transcription, DNA-templated (https://www.ebi.ac.uk/QuickGO/term/GO:0045893)
GO:0001541 : ovarian follicle development (https://www.ebi.ac.uk/QuickGO/term/GO:0001541)
GO:0030509 : BMP signaling pathway (https://www.ebi.ac.uk/QuickGO/term/GO:0030509)
GO:0048468 : cell development (https://www.ebi.ac.uk/QuickGO/term/GO:0048468)
GO:0060016 : granulosa cell development (https://www.ebi.ac.uk/QuickGO/term/GO:0060016)
GO:0010862 : positive regulation of pathway-restricted SMAD protein phosphorylation (https://www.ebi.ac.uk/QuickGO/term/GO:0010862)
GO:0042981 : regulation of apoptotic process (https://www.ebi.ac.uk/QuickGO/term/GO:0042981)
GO:0043408 : regulation of MAPK cascade (https://www.ebi.ac.uk/QuickGO/term/GO:0043408)
GO:0060395 : SMAD protein signal transduction (https://www.ebi.ac.uk/QuickGO/term/GO:0060395)

GO - Cellular Component

GO:0005737 : cytoplasm (https://www.ebi.ac.uk/QuickGO/term/GO:0005737)
GO:0005615 : extracellular space (https://www.ebi.ac.uk/QuickGO/term/GO:0005615)

Presumptive Null

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title)

Molecular Type

Coding (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Coding^#gephebase-summary-title)

Aberration Type

SNP (https://www.gephebase.org/search-criteria?/and+Aberration Type=^SNP^#gephebase-summary-title)

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

c.1100G>T p.S367I

Experimental Evidence

Candidate Gene (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Candidate Gene^#gephebase-summary-title)

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Ser	Ile	367

Main Reference

Mutations in the genes for oocyte-derived growth factors GDF9 and BMP15 are associated with both increased ovulation rate and sterility in Cambridge and Belclare sheep (Ovis aries). (2004) (https://pubmed.ncbi.nlm.nih.gov/14627550)

Authors

Hanrahan JP; Gregan SM; Mulsant P; Mullen M; Davis GH; Powell R; Galloway SM

Abstract

Belclare and Cambridge are prolific sheep breeds, the origins of which involved selecting ewes with exceptionally high litter size records from commercial flocks. The variation in ovulation rate in both breeds is consistent with segregation of a gene (or genes) with a large effect on this trait. Sterile ewes, due to a failure of normal ovarian follicle development, occur in both breeds. New naturally occurring mutations in genes for the oocyte-derived growth factors growth differentiation factor 9 (GDF9) and bone morphogenetic protein 15 (BMP15) are described. These mutations are associated with increased ovulation rate in heterozygous carriers and sterility in homozygous carriers in both breeds. This is the first time that a mutation in the gene for GDF9 has been found that causes increased ovulation rate and infertility in a manner similar to inactivating mutations in BMP15, and shows that GDF9 is essential for normal folliculogenesis in sheep. Furthermore, it is shown, for the first time in any species, that individuals with mutations in both GDF9 and BMP15 have a greater ovulation rate than sheep with either of the mutations separately.

Additional References

RELATED GEPHE

Related Genes

3 (B4GALNT2, BMP receptor IB (BMPRIB), GDF9) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9940^/and+Trait=Fertility/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

9 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^BMP15^/and+Taxon ID=^9940^/or+Gene Gephebase=^BMP15^/and+Taxon ID=^9940^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

Heterozygote shows phenotype; homozygote results in ovarian failure ; @HeterozygoteAdvantage @SexualTrait https://omia.org/OMIA001719/9940/